

Primary Author	Year	Journal	First aa Affected	Protein Change	Description	cDNA Change	Clinical Details
Cronin	2009	Pig Cell Mel Res	43	p.Arg43Gln		c.G128A	Primary melanoma
Cronin	2009	Pig Cell Mel Res	361	p.Ala361Val		c.C1082T	Primary melanoma
Cronin	2009	Pig Cell Mel Res	413	p.Gly413Ser		c.G1237A	Primary melanoma
Cronin	2009	Pig Cell Mel Res	413	p.Gly413Asp		c.G1238A	Primary melanoma
Cronin	2009	Pig Cell Mel Res	414	p.His414Tyr		c.C1240T	Primary melanoma
Cronin	2009	Pig Cell Mel Res	424	p.Ala424Val		c.C1271T	Primary melanoma
Cronin	2009	Pig Cell Mel Res	15	p.Val15AlafsX11		c.44_62delTGGGCTCG GAGGAGCCCCG	Metastatic melanoma
Cronin	2009	Pig Cell Mel Res	125	p.Gln125X		c.C373T	Metastatic melanoma
Cronin	2009	Pig Cell Mel Res	451	p.Ser451ThrfsX67		c.1352_1359delGCCCG ACA	Metastatic melanoma
Iso	2008	AJMG	169	p.Pro169ArgfsX117		c.506delC	WS2
Chen	2010	Biochem Biophys Res Comm	38	p.Gly38AlafsX71		c.113delG	WS2
Chen	2010	Biochem Biophys Res Comm	38	p.Gly38ArgfsX58		c.110_219del110	WS2
Chen	2010	Biochem Biophys Res Comm	43	p.Arg43X		c.126_127delGCinsTT	WS2
Pingault	2010	Hum Mutation	17	p.Ser17CysfsX7		c.50_73delinsGCCCGA CGCTAGGGCCCTAG	WS4
Sanchez-Mejias	2010	J Mol Med	38	p.Gly38GlnfsX21		c.112_131delGGCGGA TCGGGCCTGCGAGC	WS4
Pingault	2002	Hum Genet	43	p.Arg43X		c.126_127delGCinsCT	WS4
Sham	2001	J Med Genet	57	p.Glu57SerfsX52		c.169delG	WS4
Pingault	1998	Nat Genet	83	p.Tyr83X		c.249C>A	WS4
Jiang	2011	Biochem Biophys Res Comm	85	p.Trp85X		c.254G>A	WS4
Pingault	2002	Hum Genet	110	p.Ala110LeufsX23		c.328_329delGC	WS4
Morin	2008	AJMG	157	p.Ala157Val		c.470C>T	WS4
Pingault	1998	Nat Genet	160	p.Leu160_Arg161dup		c.477_482dupGCTCCG	WS4
Pingault	2010	Hum Mutation	173	P.Tyr173X		c.519C>G	WS4, with no hypopigmentation
Pingault	1998	Nat Genet	189	p.Glu189X		c.565G>T	WS4
Southard-Smith	1999	Genome Res	207	p.Tyr207X		c.621C>A or G?	WS4
Pingault	2010	Hum Mutation	215	p.Arg215ProfsX64		c.644_648delGGCAC	WS4
Jiang	2011	Biochem Biophys Res Comm	234	Intron splice mutation; predicted fs and truncation after 46aa addition		c.698-2A>T	WS4
Pingault	2002	Hum Genet	261	p.Arg261AlafsX25		c.780delG	WS4
Shimotake	2007	Ped Surg	261	p.Arg261AlafsX25		c.780delG	WS4
Pingault	2010	Hum Mutation	271	p.Ile271SerfsX15		c.811delA	WS4
Pingault	2010	Hum Mutation	350	p.Val350CysfsX52		c.1047dupT	WS4
Pingault	1998	Nat Genet	359	p.Glu359AspfsX42		c.1077_1078delGA	WS4
Toki	2003	Pediatr Surg Int	376	p.Ser376X		Unpublished	WS4
Pingault	2010	Hum Mutation	399	p.Gln399ValfsX2		c.1195_1196delCA	WS4, with no hypopigmentation
Sanchez-Mejias	2010	J Mol Med	52	p.Gly52AlafsX56	DNA change confirmed with author	c.155delG	HSCR
Bondurand	1999	Hum Mol Genet	135	p.Ser135Thr		c.404G>C	mild YDBS
Barnett	2009	AJMG	174	p.Gln174Pro		c.521A>C	PCW, with no HSCR; hypo- and hyperpigmentation, cochlear/olfactory nerve aplasia
Sznajer	2008	AJMG	234	Intron splice mutation; predicted fs and truncation after 46aa addition		c.698-2A>C	PCW, but no HSCR
Pingault	2002	Hum Genet	234	p.Gln234X		c.700C>T	PCWH
Inoue	2002	Ann Neurol	250	p.Gln250X		c.748C>T	PCWH
Touraine	2000	Am J Hum Genet	251	p.Ser251X		c.752C>A	PCWH
Pingault	2000	Ann Neurol	266	p.Gly266AlafsX20		c.797delG	PCWH
Inoue	2004	Nat Genet	283	p.His283LeufsX11		c.847_848insT	PCWH
Unzicker	2011	Eu J Ped	293	p.Asp293GlyfsX10		c.850_877dup28	PCWH
Vinuela	2009	Am J Med Genet	306	p.His306ThrfssX5		c.915delG	PCWH
Pingault	2010	Hum Mutation	308	p.Gly308AlafsX3		c.921delA	PCWH
Inoue	2004	Nat Genet	313	p.Tyr313X		c.939C>G	PCWH
Inoue	2004	Nat Genet	313	p.Tyr313X		c.938dupA	PCWH
Touraine	2000	Am J Hum Genet	313	p.Tyr313X		c.939C>A	PCWH
Touraine	2000	Am J Hum Genet	313	p.Tyr313X		c.939C>A	PCWH
Verheij	2006	Eur J Ped Neur	346	p.Ser346X	Corrected per Pingault, 2010	c.1037C>G	PCWH
Inoue	2004	Nat Genet	364	p.Gln364X		c.1090C>T	PCWH

Pingault	2002	Hum Genet	372	p.Gln372X		c.1114C>T	PCWH
Southard-Smith	1999	Genome Res	377	p.Gln377X		Unpublished	PCWH
Inoue	1999	Ann Neurol	467	p.X467CysextX82		c.1400_1411delAAAG GGGCCCT	PCWH
Pingault	2010	Hum Mutation	467	p.X467TyrextX86		c.1401A>C	PCWH
Sham	2001	J Med Genet	467	p.X467LysextX86		c.1399T>A	PCWH
Girard	2006	FEBS Letters	55	Lys55	Sumoylation site		
Girard	2006	FEBS Letters	246	Lys246	Sumoylation site		
Girard	2006	FEBS Letters	357	Lys357	Sumoylation site		
Iwamoto	2006	AJMG B	6	p.Asp6	Synonomous polymorphism	c.18C>T(p.=)	Detected in HSCR; 4.95% allele frequency in controls
Bondurand	2007	Am J Hum Genet	92	p.Val92Leu	Missense polymorphism	c.274G>C	PCWH patient with SOX10 deletion harbored this variant in non-deleted allele; variant has normal protein function in vitro
rs61756177			201	p.Ala201Thr	Missense polymorphism	c.601G>A	
rs17850220			226	p.Gly226	Synonomous polymorphism	c.678G>T(p.=)	
Sanchez-Mejias	2010	J Mol Med	228	p.Pro228	Synonomous polymorphism	c.684C>T(p.=)	Detected in HSCR; not seen in controls
rs5756870			255	p.Asp255Asn	Missense polymorphism	c.763G>A	
rs139884			309	p.His309	Synonomous polymorphism	c.927T>C(p.=)	0.402 allele heterozygosity
Maeno	2007	Psych Gen	309	p.His309	Synonomous polymorphism	c.927T>C(p.=)	Schizophrenia in Japanese
Sanchez-Mejias	2010	J Mol Med	419	p.Ser419	Synonomous polymorphism	c.1257T>C(p.=)	Detected in HSCR; not seen in controls
rs6000966			430	p.P430P	Synonomous polymorphism	c.1290C>A(p.=)	
rs74718340			455	p.Trp455Gly	Missense polymorphism	c.1363T>G	
Schreiner	2007	Development	71	orthologous to p.Cys71_Arg73delins	Mouse model; <i>tm4Weg</i>		
Southard-Smith	1998	Nat Genet	194	orthologous to p.Glu194GlyfsX99	Mouse model; <i>Dom</i>	mouse c.929insG	
Schreiner	2007	Development	233	orthologous to p.Gly233_His306del	Mouse model; <i>tm5Weg</i>		
www.informatics.jax.org	2010	Online submission	131	orthologous to p.Asn131Lys	Mouse model; <i>Dal</i>	mouse c.676T>G	
Dutton	2001	Development	82	p.Gly82fsX9	Zebrafish model; <i>t3</i>		
Carney	2006	J Biol Chem	113	p.Val113Met	Zebrafish model; <i>baz1</i>		
Dutton	2001	Development	138	p.Leu138Gln	Zebrafish model; <i>m618</i>		
Dutton	2001	Development	353	p.Lys353X	Zebrafish model; <i>tw2</i> and <i>tw11</i>		

Abbreviations:

p = protein
c = cDNA
X = stop

fs = frameshift

dup = duplication

ins = insertion

ext = extension

WS4 = Waardenburg syndrome 4

PCWH = peripheral demyelinating neuropathy, central dysmyelination, Waardenburg syndrome, and Hirschsprung disease

HSCR = Hirschsprung disease

YDBS = Yemenite deaf-blind hypopigmentation syndrome

WS2 = Waardenburg syndrome 2